Management of children with Epidermolysis Bullosa: Therapeutic education for parents and carers

European Reference Networks | SKIN

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Indice

Purpose of this booklet

Hereditary epidermolysis bullosa (EB) has a major impact on the life of patients, family members and healthcare professionals, with significant emotional and physical challenges. Day-to-day care requires enormous commitment from the patient and family members.

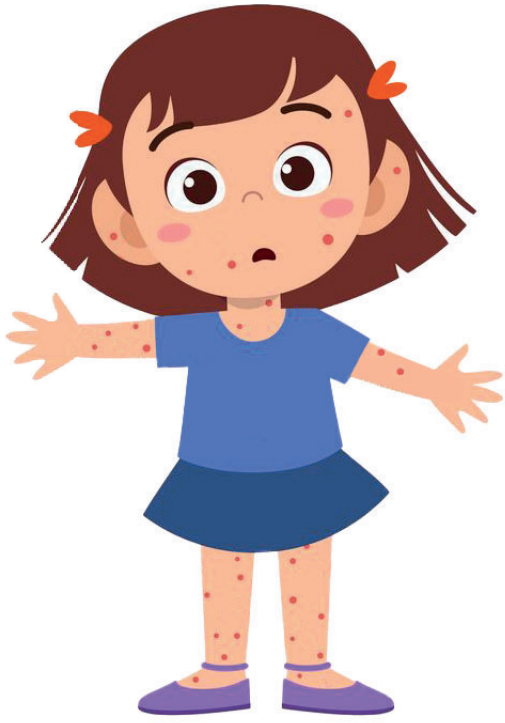
This therapeutic education manual aims to empower families to manage the condition themselves while maintaining regular contact and follow-up by the EB clinic.

This handbook, validated by the European Network for Rare Skin Diseases (ERN-Skin), is designed to guide, clarify and to offer specialised support to patients, their families and healthcare professionals by providing up-to-date guidance and advice on the daily care of patients with EB.

Introduction

Epidermolysis bullosa (EB) is a large group of genetic skin diseases. There are many different types but the common factor is skin fragility with blistering after minimal trauma.

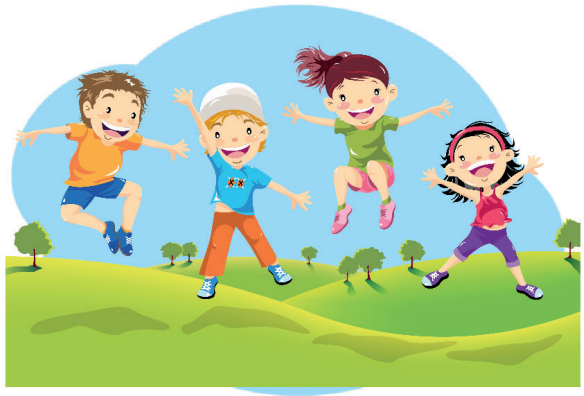
The most recent classification distinguishes four major groups: EB simplex (EBS), junctional EB (JEB), dystrophic EB (DEB) and Kindler EB (KEB).



The diagnosis of EB subtype should be established as early as possible. It requires a small skin sample (biopsy) which is examined by different techniques: standard light microscopy can exclude other diseases; a more specifics technique called immunofluorescence antigen mapping shows which skin component is reduced or absent; transmission electron microscopy, if available, can be useful for the diagnosis of certain EB subtypes. A blood sample should also be taken from the patient and parents for genetic testing in order to confirm the diagnosis, enable genetic counselling and if necessary to allow prenatal diagnosis.[2]

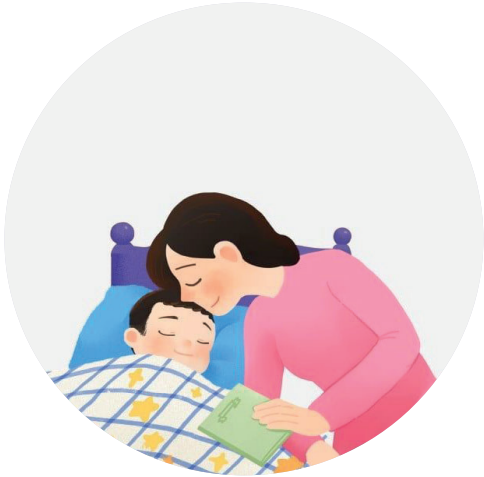
The presence of multiple, painful wounds, as well as their persistence and recurrence, makes treatment complicated.

There is still no cure for EB, although numerous studies and research projects are underway. However, good daily management can improve quality of life and prevent and/or delay complications of the disease.



Care of EB newborn and infant

Essentially, babies with EB should be treated like healthy babies, but of course specific measures and extra help are required.



To lift the baby without traumatising the skin, put one hand behind the neck and head and the other beneath the buttocks, let the baby roll back towards you and then lift him or her.

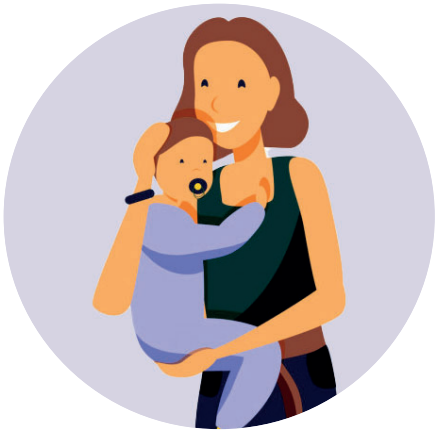
Avoid lifting the baby by holding under the armpits because this can cause trauma and blistering. Another technique is to lie the baby on a pillow, mattress, sheet or advanced foam dressing and to lift the child by sliding your hands below the mattress or sheet.

Breastfeeding should be encouraged for two main reasons:

- Breast milk provides important nutrients, which strengthen the baby's immunity.
- Close physical contact is beneficial for both mother and baby (kangaroo care).

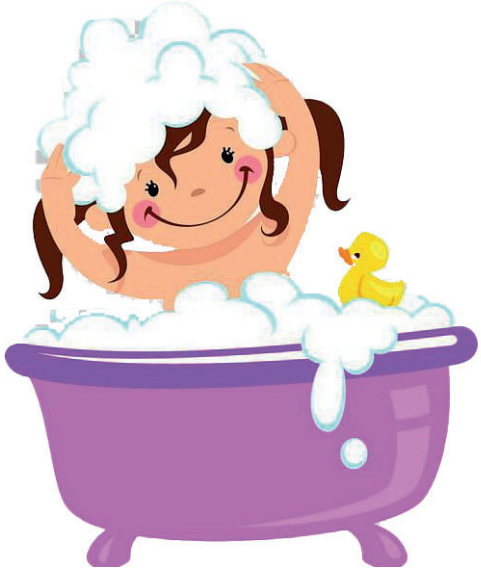
Before feeding, apply a lubricant such as soft paraffin to the mother's nipples and the baby's face. If the baby has difficulty sucking, extract the milk and give it in a bottle with a teat softened in warm water or of soft silicone, or a Haberman feeder, which has a long teat to reduce sucking and trauma to the nose. It is also possible to enlarge the hole of the teat to reduce sucking effort. Alternatively, put the milk into a cup and administer it with a soft silicone spoon. Avoid straws absolutely.

A pediatrician should also provide regular follow-up of the patient.



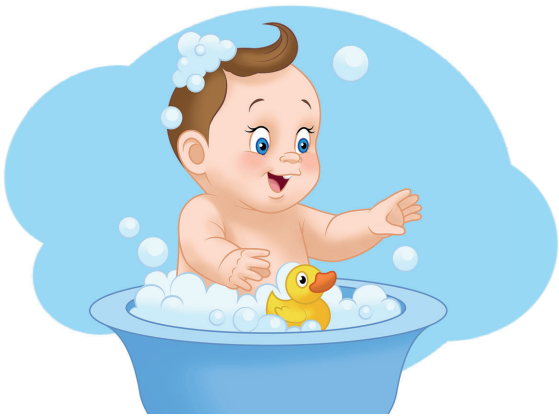
Bathing

Bathing is very important for the following reasons: (i) it cleanses and reduces the risk of infection; (ii) it removes crusts and scales; (iii) it reduces itching; and (iv) it allows all lesions to be checked. However, it can be painful so the prescribed analgesic must be given beforehand. Bathing needs a quiet, well-lit environment with suitable entertainment available to calm and distract the child.



Bathing frequency depends on the types of lesion and dressings used. Daily bathing is recommended if there are infected wounds or when dressings are stuck to the skin. Before bathing, prepare everything you will need in a clean area: soft towel, emollient/ oil based cleanser (for dry skin and hyperkeratotic lesions) or mild antiseptic cleanser (for widespread skin infection), and dressings.

In addition, line the inside of the bathtub with foam or a soft towel. Check the water temperature. Remove the outer dressings and place the child in the tub with the primary dressings still in place so they can be gently soaked off.



Use a soft towel and pat the child dry to avoid blistering and proceed with dressing.

Dressings

General principles

Blisters occur following minor trauma and during hot weather. They are recurrent and can turn into painful sores and chronic ulcers.
Dressings are therefore essential to reduce symptoms (pain and itching), and to prevent skin and internal complications (e.g., infection, chronicity, scarring, contractures, malnutrition and the development of skin cancer).

Therefore it is worth putting as much effort as possible into skin care. The effort involved for the patient and family is more than compensated for by the patient's well-being and improved health.

Parents/carers should learn about wound management at the EB centre.
Dressings are painful and prescribed analgesic should be given before any procedure.
The frequency of dressings varies according to the type of wounds and dressing used: your EB specialist will advise.

Adhesive dressings and tapes must be avoided.

Frequent use of emollients is recommended to moisturize the skin, remove the scales, reduce itching, and prevent thick hyperkeratosis which may mask chronic wounds and delay the diagnosis of possible squamous cell carcinoma (a complication in some EB subtypes).

Palms and soles should be checked for hyperkeratosis which can cause pain and difficulty with walking. This can be treated with keratolytic ointment which softens thickened skin.

Do not hesitate to consult the doctor or nurse at the EB clinic if you are worried.

1. Wash your hands thoroughly before starting.

2. Prepare all necessary dressing materials on a clean surface, cutting dressings as needed.

3. Don't rush the dressings and check frequently whether the patient is upset, uncomfortable or in pain.

4. Administer analgesic therapy as directed to avoid patient suffering.

5. Create a pleasant environment suitable for the age and wishes of the patient (music, video, game, chat...).

6. Make sure the patient's position is comfortable with adequate protection for areas susceptible to pressure before the person lies down or sits down (back and buttocks).

7. Carefully observe the skin and all lesions when removing dressings to check wound bed, odour, amount of secretions, edges of the wounds, and hot temperature of affected area. Ask about the duration of the wounds.

8. The choice of topical agents and dressings depends on the type of lesion, not the type of EB, and must take into account the efficacy and safety of the product, patient acceptance and cost.

9. Clean the lesions as advised by the EB specialist, patting gently.

10. Lance the blisters, using a sterile disposable needle or finger prick lancet, and drain. The blister roof should be left in place to improve re-epithelisation and to reduce infection risk and pain.

11. Apply topical agents as prescribed by the EB specialist and according to the type and location of the lesion.

12. Moisturise dry no-blistered skin.

13. Dressings should be fixed with tubular bandages of various sizes according to the affected body area, making sure they do not restrict the circulation or movement. Adhesive tapes must not be applied to skin.

14. In some EB subtypes, dress the fingers and toes by separating them from each other to prevent or delay pseudosyndactyly (fusion).

15. For patients with widespread skin damage, antimicrobial silk garments may be of great help.

Specific recommendations

Dry or lightly exuding lesions:

- Polyamide dressings with a soft silicone layer or with a lipid-colloid contact layer, changed every 2-4 days.
- Thin polyurethane foam dressings with a soft silicone layer, changed every 2-4 days.
- Hydrogel dressings should be changed daily.
- Dressings can be applied directly to the wound or after application of topical antimicrobial products (as prescribed by the EB specialist).
- Cover the dressing with soft material suitable for EB and for the lesion site. The same material can also be used to protect healthy skin at sites of trauma.

Exuding or heavy exuding lesions:

- Polyamide dressings with a soft silicone layer on both sides.
- Thin, super absorbent polyurethane foam dressings with soft silicone contact layer.
- Dressings composed of hydrogels or alginates that require a secondary dressing of foam or hydrocolloids.
- Gelling hydrofibres.
- Polymeric membranes.
- Concurrent use of topical antimicrobial products if advised by the EB specialist.

Critically colonized and infected wounds:

- A swab should be sent to the laboratory but antibiotic treatment should be started while awaiting the result of culture and sensitivity testing.
- Clean the wound with antiseptics.
- Apply topical antimicrobials and suitable topical antibiotics avoiding systemic antibiotics where possible.
- Creams and dressings containing silver are indicated in both types of wounds. However, the potential toxicity of silver should be taken into account, and its use over large areas for long periods and in young children should be avoided.
- The same dressings are used for heavily exuding wounds and should be changed daily.

Hyperkeratotic and/or crusted lesions:

- Bath or soak the affected area at least two to three times a week.
- Gently remove the thick scale and/or crust after repeated application of moisturising agents or vaseline.
- Apply emollient creams regularly to prevent recurrent scaling or hyperkeratosis. possible.

Wounds with exuberant granulation tissue:

- Short courses of topical corticosteroids or silver with constant monitoring by the caregiver to avoid side effects.

How to recognize an infected wound?

Signs of infection are:

- enlargement of wounds and red edges.
- gauze more soiled than usual due to increased secretions.
- change in colour of secretions.
- foul smell.
- increased pain/itching.

When infected wounds are multiple and/or extensive, the patient may be febrile. In this case, the EB specialist should be consulted immediately, a systemic antibiotic may be required and dressings should be changed daily.

Management of chronic wounds

A wound that takes longer time to heal than others despite appropriate dressings is considered chronic.

Such wounds are sometimes masked by thick crusts. It is important to be aware of these lesions, to prevent them, to clean them properly removing scales and crusts and to monitor them because in some EB subtypes, they are at risk over time of skin cancer.

If skin cancer is suspected the dermatologist must be consulted as soon as possible.

Skin hydration

It is important to keep the skin well hydrated by following the EB specialist's instructions.

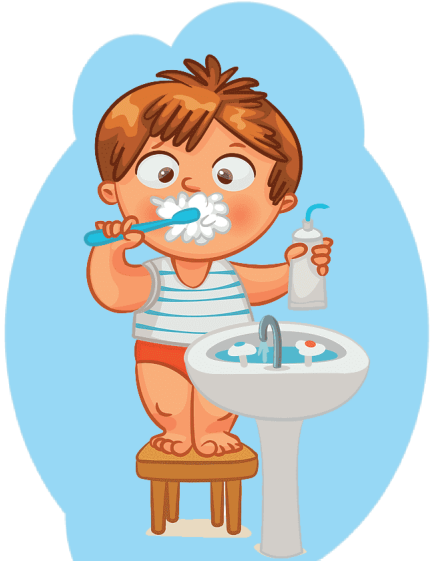
Dry skin causes and/or worsens itching, which in turn leads to scratching, more EB lesions and more suffering. Obviously, moisturising alone may not control itching, but it is an important support.

Mouth care

The oral mucosa is affected in many EB subtypes.From the age of 3-6 months, the child should visit a dentist and oral hygienist regularly, to familiarise them with oral examination. Regular dental follow-up is essential from the earliest months with variable frequency, to be agreed by the dentist and the EB specialist.

The oral mucosa is affected in many EB subtypes, the following are recommended:

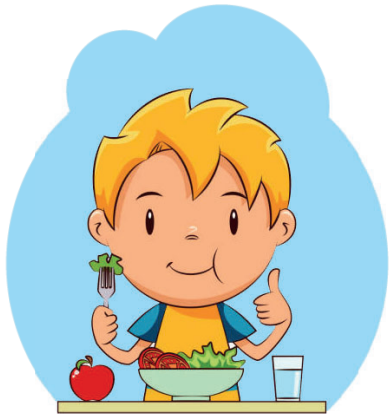
- a cotton ball, cotton swab or glove to clean sensitive gums and teeth.
- a soft-bristled toothbrush.
- a toothbrush with a small head for patients with microstomia (small mouth); the handle may need adaptations if the child's fingers are fused.
- toothpaste of appropriate taste for children to encourage tooth cleaning.
- low-sugar diets to reduce the risk of caries.



- fluoride-based topical agents.
- antiseptic and topical agents as prescribed by the EB specialist.

Nutrition and food intake

Blisters and wounds of the oral cavity and oesophagus cause pain and often become a reason for refusing to eat. Thus, it is important to provide soft food, taking into account the quality and quantity in order to ensure adequate nutritional intake.



The patient should be regularly followed up by a dietician and nutritionist to optimise nutritional status and growth. Recommendations include:

- Include the patient in family meals from childhood.
- Provide multivitamins and food supplements to increase calorie intake.
- Give plenty of fluids.
- Avoid excess salt and spicy seasoning which can irritate the oral mucosa.
- Minimise sweets to prevent dental caries.

For patients with significant involvement of the mouth and/or oesophagus:

- Do not offer hard, crunchy food.
- Suggest pureed food.
- Offer small and frequent meals.
- Consult the EB specialist if there are persisting mouth ulcers, or difficulty with swallowing.

Clothing

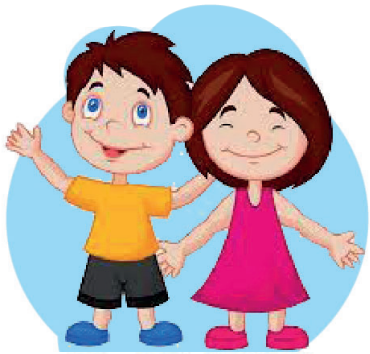
Clothing should be comfortable and soft. Rough seams can cause friction so undergarments can be worn inside out.

Remove or avoid labels, zippers, and elastic that can rub the skin.

Choose clothing that is easy to put on and remove, without chafing on the head, ears and armpits.

Disposable nappies should be the right size. A silicone barrier cream under the edges of the nappies is recommended to protect the skin.

Choose soft cotton material for bedding.



Footwear

It must always be remembered that immobilisation promotes joint contractures and early osteoporosis. It is very important to consider comfort so that the patient can move properly, play, and walk.

Soft, seamless, easy-to-wear socks and shoes are recommended; shoes size should take into account dressings if necessary.

Some patients require shoes with specific additional cushioning (e.g. foam insoles).

Vaccination

EB is not a controindication to vaccination. Immunization schedules for infectious diseases should be followed as normal, and chicken pox vaccination is recommended. Obviously, specific care must be taken during the procedure:

- The doctor should be informed about skin fragility.
- Antiseptic should be applied gently to prevent blistering.
- The vaccine must be carried out on uninflamed skin.
- After inoculation, no adhesive dressing should be applied to the skin.

Sporting activities

EB patients are encouraged to exercise, and should be informed about suitable choices of sports. The benefits of sports are:

- Fun, relaxation, distraction, and socialising.
- Promoting mobilisation and reduced risk of osteoporosis (frequent complication in some EB subtypes).
- Not feeling different from peers.

However, certain precautions are necessary, particularly for patients with some EB subtypes:

- Avoid traumatic or contact sports.
- Direct the child from an early age towards sports that encourage movement.
- Change the dressings if the activity has caused sweating, even it was not planned for that day.

Psychological support

Psychological support should be offered by the EB centre to:

- The parents of EB newborn as soon as they are given the diagnosis.
- The patient as soon as they become aware of the disease.
- The healthcare providers to improve empathy and relationships with the patient/ caregivers.

Psychological support should aim at supporting the patient and the family in managing daily life, school and social integration, play activities, etc. It must therefore be appropriate to the needs of each patient and family and regularly adapted to changes in circumstances and age.

Psychological support is a positive intervention allowing confront issues, overcome difficulties and move forwards.

If it is not helping, consider whether another psychologist might be better suited to this particular situation.

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Revised by the members of epidermolysis bullosa sub-thematic group in ERN-Skin

Validated and approved by Prof. C. Bodemer Coordinator of ERN-Skin

The aim of this small handbook is to provide a short, practical tool for patients with epidermolysis bullosa and their families for daily disease management. Obviously, this brochure does not in any way replace suggestions and follow-up plan prescribed at the EB treatment centre!

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